Potential role for a postmortem echocardiographic evaluation in infants with suspected dilated cardiomyopathy

Mani Ram Krishna[1] and Usha Nandhini Sennaiyan[1]

[1]Tiny Hearts Fetal and Pediatric Cardiac Clinic Thanjavur

October 26, 2022

Abstract

Dilated cardiomyopathy in infants could potentially be secondary to structural heart diseases such as LV outflow obstruction and ALCAPA. These conditions can be diagnosed by a dedicated echocardiogram. When such an evaluation was not possible, a postmortem echocardiogram could potentially provide clues about the possibility of a structural heart disease

Potential role for a postmortem echocardiographic evaluation in infants with suspected dilated cardiomyopathy

Mani Ram Krishna and Usha Nandhini Sennaiyan

Tiny Hearts Fetal and Pediatric Cardiac Clinic

Thanjavur

Keywords: postmortem echocardiogram, sudden infant death, ALCAPA

Correspondence to

Dr Mani Ram Krishna

Tiny Hearts Fetal and Pediatric Cardiac Clinic

No 7, V.O.C. Nagar,

Thanjavur – 613007

E-mail: mann_comp@hotmail.com

Conflict of Interest: None

Funding involved in the study: None

Data Availability Statement:

The data that support the findings of this study are available on request from the corresponding author. The data are not publicly available due to privacy or ethical restrictions.

Dilated cardiomyopathy in infants is often secondary to reversible causes(1). These include structural heart diseases such as aortic stenosis, coarctation of aorta and anomalous origin of left coronary artery from pulmonary artery, incessant arrhythmias such as atrial tachycardia and permanent junctional reciprocating tachycardia as well as nutritional causes such as Vitamin B1 and calcium deficiency(2). Anomalous origin of the left coronary artery from the pulmonary artery is an important reversible cause of left ventricular (LV) dysfunction in infants(3).

A 6-month-old baby was referred to us by a pediatrician for a suspected dilated cardiomyopathy. The baby had presented with feeding difficulty, fast breathing and poor weight gain over three months with an acute deterioration over the previous week. A chest x-ray at the referring hospital had demonstrated cardiomegaly and a screening echocardiogram had demonstrated severe left ventricular dysfunction. An electrocardiogram had not been obtained. The baby suffered a cardiac arrest in the ambulance enroute to our center. She was intubated and an inter-osseous access was obtained. Despite high quality cardiopulmonary resuscitation for 30 minutes, the baby could not be resuscitated and was declared dead. The cause for left ventricular dysfunction could not be established. We hence performed a postmortem echocardiogram. There was no cardiac activity, and the left ventricle was dilated and filled with thrombus. The imaging of the left ventricle in the para-sternal long axis did not suggest important aortic obstruction with a normal sized aortic annulus and ascending aorta (Figure 1). The imaging of the aortic valve in the para-sternal short axis view suggested that the origin of the left coronary artery (Figure 2A and Video 1) and the right coronary artery (Figure 2B) were normal although this could not be inconclusively proven by color Doppler imaging. The parents refused a postmortem evaluation. However, after detailed counseling, they agreed for genetic testing. Whole exome sequencing of deoxyribonucleic acid extracted from peripheral blood revealed a homozygous variant in the ALMS 1 gene (c.12080_12083delTACT) (Transcript ID NM_015120.4) causative of Alstrom syndrome. Dilated cardiomyopathy is a well-recognized presentation of Alstrom syndrome in infancy.

In the past, it has been suggested that a postmortem echocardiogram could be used as a guide to direct the potential need for postmortem examination in sudden infant death and sudden unexpected deaths in the young(4). Although our findings could not be backed by postmortem anatomical confirmation, the images appeared to be reasonably convincing to suggest that a postmortem echocardiogram can be considered for infants in whom a comprehensive cardiac evaluation could not be performed before death.

Compliance with ethical standards:

Funding: The authors did not receive any support from any organisation for this work

Competing interests: The authors have no relevant financial or non-financial interests to disclose

Informed Consent: Consent was obtained from the parents of the infant

Legends

Figure 1: Post-mortem echocardiogram in a para-sternal long axis view demonstrating the left ventricular outflow tract (LV – left ventricle)

Figure 1A and Video 1: Post-mortem echocardiogram in a para-sternal short axis view demonstrating what appears to be the left coronary artery (green arrow in Figure 1A) arising from the left coronary sinus of the aorta

Figure 2B: Post-mortem echocardiogram in a para-sternal short axis view demonstrating the likely origin of the right coronary artery (RCA) from the right coronary sinus of the aorta

References

1. Lipshultz SE, Law YM, Asante-Korang A, Austin ED, Dipchand AI, Everitt MD, et al. Cardiomyopathy in Children: Classification and Diagnosis: A Scientific Statement From the American Heart Association. Circulation. 2019;140(1):e9-e68.

2. Kumar RK. A practical approach for the diagnosis and management of dilated cardiomyopathy. Indian J Pediatr. 2002;69(4):341-50.

3. Krishna MR. ALCAPA-A Reversible Cause of Dilated Cardiomyopathy in Infants. Indian J Pediatr. 2021.

4. Ker J, Toit-Prinsloo LD, Van Heerden W, Saayman G. Post-mortem echocardiography as a guide to cardiac autopsy-a worthwhile concept? Clin Med Insights Cardiol. 2010;4:59-61.